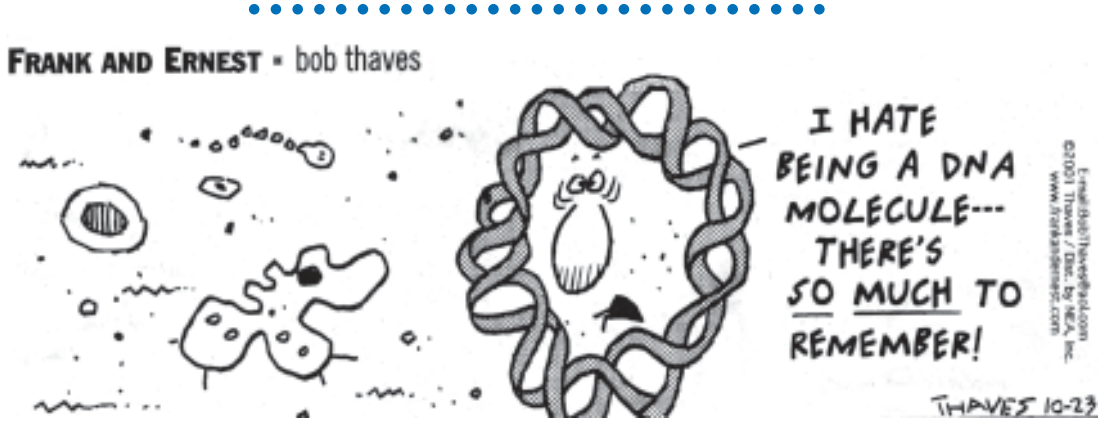
Hawai`i Department of Health
Genetics Program
741 Sunset Avenue
Honolulu, HI 96816

Genetics Causes of Congenital Hearing Loss

Approximately, 1 in 300 babies is born with a hearing loss. While most cases of hearing loss in infants are believed to have a genetic cause, more research is needed to better define all causes.

We are excited to report the launch of a research project in Hawaii (in conjunction with three other states) that will look at the genetic causes of hearing loss in infants. All infants diagnosed with permanent hearing loss through newborn hearing screening or before their third birthday will be invited to join the study. Participation in the study will include a full genetics evaluation, discussion with a genetic counselor and geneticist (a doctor who specializes in genetics), and genetic testing. Parents will be contacted with the testing results. Parents will also have the option to have part of their child's blood sample stored for future tests as they become available. For more information, please contact Allison Taylor (733-4998) or Lianne Hasegawa (733-9039).



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GENE NEWS

A Publication of the Hawai`i Department of Health Genetics Program

New Legislation

The 2002 Hawaii State Legislature passed two genetics related bills and both were signed into law by Governor Ben Cayetano. Thank you to the many individuals and organizations for your support.

Genetics Anti-Discrimination for Employment (Act 217)
Hawai`i has become the 28th state to specifically prohibit the use of genetic information or genetic testing for employment purposes (Act 217). This law applies to potential employees, as well as current employees.

Hawai`i Birth Defects Program (Act 252)
The program was initially established in 1988 as a research project. After 14 years of collecting information about birth defects in Hawai`i, the program is now officially mandated to be part of the Department of Health and will be self sustaining. For further information, please contact Ruth Merz at 587-4120 or hbdp@crch.hawaii.edu.

Attention Health Care Providers!

The Genetics Program is developing a Spring 2003 conference for Health Care providers. The conference will cover the issues physicians rated highest among interest levels during the 2000-2001 physician needs assessment including: breast cancer, colon cancer, diabetes, and cardiovascular disease. More information will be in the next GeneNews issue.

New faces

The State Genetics Program welcomes two genetic counselors to the program: Allison Taylor and Lianne Hasegawa. Allison joins us as the Project Coordinator for the Gene AID project, and Lianne is a Project Specialist for the Tandem Mass Spectrometry Project.

From left to right:
Allison Taylor,
Nicole Sameit
Sharon Hirose (front)
Lianne Hasegawa
Sylvia Au

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Hawai`i Department of Health

Hawai'i Genetics Program

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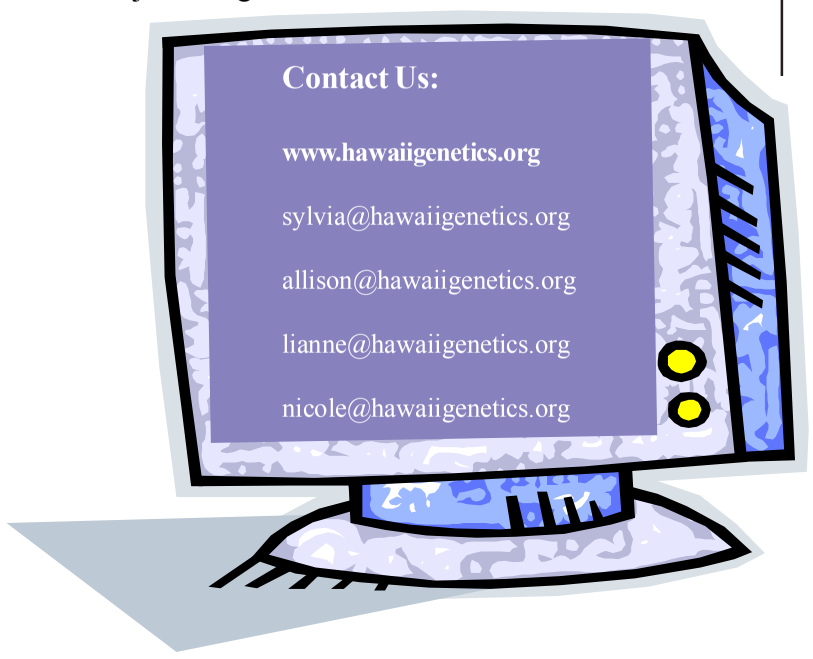
State Plan Update

We are in the final stages of the Hawaii Genetic Assessment and Planning Project (HGAPP). The draft revision of the State Genetics Plan is available for public review and comment at www.hawaiigenetics.org.

The revised Plan is an update of the original 1993 State Genetics Plan and will incorporate the recommendations and findings from our recent statewide genetics needs assessments. The responses from the needs assessment have enabled us to determine the baseline level of genetic awareness of our population, the general attitudes towards genetic testing and screening, and the preferred methods and subjects for genetics education.



Conference Overview

The second conference in the series, entitled "The Impact of Genetics on Public Health: Putting the Pieces Together", was held May 7-8th, 2002 at the Hilton Hawaiian Village. The event, targeting public health professionals, was a huge success with over 100 participants from across the state.

Experts from the mainland and staff from the Hawaii Department of Health spoke on a variety of topics of state and national significance. Topics included the ABCs of genetics, the Human Genome Project, the expanded newborn metabolic screening program, updates on the state genetics program, current genetics legislation, the Health Information Portability and Accountability Act (HIPAA), the genetics of hearing loss, and the genetics of autism.

The conference is part of an ongoing effort to raise awareness of the important relationship between genetics and public health and how to incorporate genetics in public health programs. An overwhelming majority of the participants rated the conference as relevant and expressed enthusiasm about the overall experience. Please check our website for upcoming educational sessions.

For more information about the Genetics Program or any of our projects, please go to our website at:

www.hawaiigenetics.org

Alpha Thalassemia in Hawaii

Alpha Thalassemia is an inherited form of anemia. Certain types of alpha thalassemia cause chronic illness and death, which is preventable if better recognized. There are many different types of alpha thalassemia, each with its own clinical pattern and significance. Because of the mixed ethnic backgrounds of our population, these various types of alpha thalassemia are especially prevalent in Hawaii.

A research project was initiated in March of 2001 by Dr. Kelley Woodruff, the DOH Newborn Metabolic Screening Program, Queen's Medical Center, and the Oregon State Public Health Lab, supported by the Clinical Research Center. The goals of the project are to:

- Identify the different types of alpha thalassemia in the infants detected through newborn screening;
- Compile information regarding ethnic backgrounds, types of gene deletions and abnormalities; and
- Correlate the newborn screening results and the actual alpha thalassemia DNA mutations.

The clinical research phase of the project is completed and the data is being analyzed. However, there has been a continued request for services, so the Queen's Genetics Thalassemia Clinic has been established. The Thalassemia Clinic will offer further genetic testing, genetic counseling and education for families with inherited anemia. You can contact the Thalassemia Clinic at 537-7633.

Tandem Mass Spectrometry Project Update

As of March 1, 2002, the Newborn Metabolic Screening and Genetics Programs successfully launched the supplemental newborn screening pilot project using technology called Tandem Mass Spectrometry (MS/MS).

Hawaii currently screens for seven mandated disorders in newborns. MS/MS technology allows us to increase this number to over thirty disorders and still have the program maintain cost effectiveness. As part of the MS/MS pilot project, this supplemental screening is being offered to parents of all infants born at Kapiolani Medical Center between March and September 2003. The pilot project has proven to be a great success with over 1600 parents agreeing to participate in the supplemental screening to date. One of the parents was so interested in the research that she has now taken a job with us as a project assistant getting informed consent from new mothers!

How Can I Get Involved?

If you are a new parent, an expectant mother, a new mother (within the past 12 months), a health care professional, or member of the general public, we need your help!

We will be conducting **focus groups** to get your feedback on how this new technology affects or may affect you, and your experience with screening so far. Focus groups will be scheduled within the next few months. If you are interested in participating in the focus groups, or for more information about the MS/MS project, please contact Lianne Hasegawa (808) 733-9039 or Nicole Sameit (808) 733-8387.

Current Statistics for Tandem Mass Spectrometry Research Project (California and Hawaii)

January 7 - August 16, 2002

Newborns Tested:	100,589
Newborns Referred for Follow-up:	285 (0 for Hawaii)
Cases detected:	11*
Turn-around time for Hawaii specimens:	12 days from time of blood collection

* Cases detected: 3 Medium Chain Acyl-CoA Dehydro genase Deficiency (MCAD), 2 Short Chain Acyl-CoA Dehydrogenase Deficiency (SCAD), 1 Glutaric Acidemia-Type II (GA-II), 2 Methylmalonic Acidemia (MMA), and 3 possible SCAD still under investigation.